U.S. Department of Health and Human Services

Office of the National Coordinator for Health Information Technology

Personalized Healthcare
Prototype Use Case
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1.0 Preface

Use cases developed for the American Health Information Community (AHIC) are based on the priorities expressed by the AHIC workgroups. These high-level use cases focus on the needs of many individuals, organizations, and systems rather than the development of a specific software system. The use cases describe involved stakeholders, information flows, issues, and systems needs that apply to the multiple participants in these arenas.

The use cases strive to provide enough detail and context for detailed policy discussions, standards harmonization, certification considerations, and architecture specifications necessary to advance the national health information technology (HIT) agenda. These high-level use cases focus, to a significant degree, on the exchange of information between organizations and systems rather than the internal activities of a particular organization or system.

During the January 2007 AHIC meeting, nine priority areas (representing over 200 identified AHIC and AHIC workgroup detailed priorities) were discussed and considered. Three of these areas (Consumer Access to Clinical Information, Medication Management, and Quality) were prioritized and developed into the 2007 Detailed Use Cases, which were published in June 2007. The Health Information Technology Standards Panel (HITSP) Technical Committees are currently conducting harmonization work on these use cases.

The remaining six priority areas from the January 2007 AHIC meeting were updated based upon AHIC feedback and were reviewed during the July 2007 AHIC meeting. These six priority areas are now being developed into the 2008 Use Cases (Remote Monitoring, Remote Consultation, Personalized Healthcare, Consultation and Transfers of Care, Public Health Case Reporting, and Immunizations & Response Management) which will be processed in the national HIT agenda activities in 2008.

The 2008 Use Cases are being developed by the Office of the National Coordinator for Health Information Technology (ONC) with opportunities for review and feedback by interested stakeholders within both the private and public sectors. To facilitate this process, the use cases are being developed in two stages:

- The **Prototype Use Cases** describe the candidate workflows for the use case at a high level, and facilitate initial discussion with stakeholders; and
- The **Detailed Use Cases** document all of the events and actions within the use case at a detailed level.

This document is a prototype use case, which describes at a high level the actors, capabilities, and information sharing needs associated with this use case. ONC is publishing the prototype use case at an earlier stage of development in order to incorporate more substantive input from interested stakeholders into the detailed use case.

The prototype use case is divided into the following sections:

- Section 2.0, Introduction and Scope, briefly describes the priority needs identified by one or more AHIC workgroups and preliminary decisions made about the scope of the use case.
- Section 3.0, Use Case Stakeholders, briefly describes individuals and organizations which participate in activities related to the use case and its components.
- Section 4.0, Issues and Obstacles, briefly describes issues or obstacles which may need to be resolved in order to achieve the capabilities described in the use case.
- Section 5.0, Perspectives, briefly describes how the use case combines similar roles (or actors) in order to describe their common needs and activities. The roles are intended to describe functional roles rather than organizations or physical entities.
- Section 6.0, Candidate Workflows, briefly describes how various perspectives interact and exchange information within the context of a workflow. The use case workflow model provides a context for understanding the information needs and is not meant to be prescriptive.
- Appendix A, the Glossary, provides draft definitions of key concepts and terms contained in the prototype.

Also within the prototype document are specific questions for which ONC would like to receive feedback during the development process. Following receipt of feedback from interested stakeholders, ONC will develop a detailed use case, which will incorporate the feedback received, fully describe the events and activities from a variety of perspectives, and include information flow diagrams.

2.0 Introduction and Scope

In January 2007, the AHIC approved a recommendation to develop a use case addressing personalized healthcare. Personalized healthcare is a process in which healthcare providers can customize treatment and management plans for patients based on their unique genetic makeup. The personalized healthcare use case will focus on the exchange of genomic/genetic test information, family health history and the use of analytical tools in the electronic health record (EHR) to support clinical decision-making. In specific terms:

- Consumers and clinicians will benefit from the inclusion of family health history, combined with genetic testing results in EHRs. This will provide useful predictive information that can lead to preemptive actions and earlier disease detection.
- Clinicians will benefit from capabilities that link large medically related genetic datasets to relate to individual-level genetic/genomic data.
- Clinicians will be better able to manage individual patients if access to education and information providing guidance on genetic test selection, risk analysis tools, and family health history information, are made available to them.
- Genetic/genomic information can be helpful in health maintenance, prevention, and disease management, which may lead to a reduction in overall health care costs by encouraging early detection and preemption.
- Clinical care data linked to genetic information can benefit researchers as they work to transform genetic/genomic discoveries into useful clinical applications.

One of the goals of the AHIC to establish a pathway, based on common data standards, to facilitate the incorporation of interoperable, clinically useful genetic and family health history information and analytical tools into EHRs to support clinical decision-making. Family health history takes time to gather, increasing the need for interoperability. Ideally, family health history would be gathered prospectively as new events occur rather than retrospectively by interview at different encounters. Likewise, accurately recording the data from genetic tests, as well as having a complete record of all genetic tests performed for a consumer, regardless of the ordering clinician, is important. Genetic information, unlike many other laboratory test information, has lifelong significance.

This use case was developed to support the many stakeholders who are active in the development and implementation of EHRs, interfaces between personal health records (PHRs) and EHRs, and health information exchange (HIE) including those engaged in activities related to standards, interoperability, harmonization, architecture, policy development, and certification. To support this need, the Personalized Healthcare Prototype Use Case focuses on the exchange of family history and genetic testing information between stakeholders in three candidate workflows:

- **Clinical Assessment.** The family history is gathered from or by the consumer in an interoperable form to be used by clinicians. This information is accessed by clinicians for the purpose of developing a diagnostic plan.

- **Genetic Testing and Reporting.** A genetic testing laboratory receives and captures genetic/genomic test orders and any accompanying information necessary for the testing. The testing laboratory performs the tests, then develops the patient report and transmits this information back to authorized providers.

- **Clinical Management.** Clinicians utilize this new diagnostic information for management of the patients. Both clinicians and consumers have access to this information via the EHR or PHR.

This use case assumes the developing presence of electronic systems such as EHRs, PHRs, and other local or web-based solutions supporting consumers and clinicians, while recognizing the issues and obstacles associated with these assumptions. This approach helps promote the development of longer-term efforts.

Combining genetic testing information with family health history information in the EHR could facilitate improvements in medical decisions, the transition to a more preemptive medical practice, and patient participation in healthcare management.

3.0 Use Case Stakeholders

Figure 3-1. Personalized Healthcare Use Case Stakeholders Table

Stakeholder	Working Definition
Clinicians	Healthcare providers with patient care responsibilities, including physicians, advanced practice nurses, pharmacists, physician assistants, nurses, medical geneticists, genetic counselors, and other credentialed personnel involved in treating patients.
Consumers	Members of the public who may receive healthcare services. These individuals may include: caregivers, patient advocates, surrogates, family members, and other parties who may be acting for, or in support of, a patient.
Genetic Testing Laboratories	Medical laboratories which perform genetic laboratory tests ordered by genetics specialists to assess the genetic status of patients. These services may be delivered through hospitals or through free-standing entities.
Genetic/Genomic Knowledge Repositories	Organizations that maintain resources, such as online servers and databases, which provide evidence-based analysis and validated knowledge, such as information on specific genetic diseases, human genomic sequence data and references to the relevant medical literature.
Health Researchers	Organizations or individuals who use health information to conduct research.
Healthcare Entities	Organizations that are engaged in or support the delivery of healthcare. These organizations could include hospitals, ambulatory clinics, long-term care facilities, community-based healthcare organizations, employers/occupational health, school health, dental clinics, psychology clinics, care delivery organizations, and other healthcare facilities.
Healthcare Payors	Insurers, including health plans, self-insured employer plans, and third party administrators, providing healthcare benefits to enrolled members and reimbursing provider organizations. As part of this role, they provide information on eligibility and coverage for individual consumers, as well as claims-based information on consumer medication history. Case management or disease management may also be supported.
Patients	Members of the public who receive healthcare services.
Public Health Agencies (local/state/federal)	Local, state, and federal government organizations and personnel that exist to help protect and improve the health of their respective constituents.
Registries	Organized systems for the collection, storage, retrieval, analysis, and dissemination of information on individual persons to support health needs.

ONC would like to receive feedback on the draft list of stakeholders and their descriptions for this use case. Please suggest additions, deletions and/or revisions to the description of the stakeholders.

4.0 Issues and Obstacles

Realizing the full benefits of personalized healthcare will be dependent on overcoming a number of issues and obstacles in today's environment. Inherent in this use case is the premise that some of these will addressed through HIT standardization and harmonization activities, policy development, HIE networks, and other related initiatives. Genetic/genomic information can provide information on disease status, predisposition to various diseases, the risk of passing on a disease to offspring, and potentially adverse or positive responses to therapeutic actions.

Confidentiality, Privacy, Security and Data Access

- Consumer data confidentiality and privacy
    - The implementation of personalized healthcare raises issues regarding the potential use of family history and disease risk and predisposition.
    - Consumers will want confidentiality, access control and information describing who has had access to their information.
    - Secondary uses of this information need to be appropriately controlled.
    - There are re-identification risks because of the uniqueness of genetic sequence information.
- Security and data access
    - Personal health data must be appropriately secured whenever stored, transmitted, archived, or destroyed.

Family Health History Information Interoperability and Security

Family health history is central to the notion of personalized healthcare in general and genetic disease diagnosis in particular. However, in the current environment there are several issues that will need to be addressed in order to utilize this important information while maintaining interoperability and confidentiality and privacy for consumers.

- Currently, family health history is obtained by interviewing the patient and/or other related individuals. Clear standards for the specific information gathered and guidelines for standard nomenclature regarding family health history are needed to support:
    - Information exchange between clinicians or between clinician and patient via PHR or EHR.

- Information exchange between and among family members via PHRs. This data exchange, while important and necessary in many instances related to genetic/genomic information, raises significant privacy and confidentiality concerns and needs to be addressed within the context of HIPAA and other privacy rules and regulations.

- There is also a need for coding for family structure and relationships. Data included in a family health history is not always precise. Terminologies must allow coding at the level of certainty that can be obtained.

Genetic/Genomic Data Interoperability

The exchange of genetic/genomic information across systems, sites and settings of care is constrained by the lack of agreed upon standards for results and nomenclature. Some specific issues are as follows:

- Limited EHR and PHR penetration.

- Fragmented nomenclature for ordering and reporting of genetic testing results.

- Lack of standards for information transferred to and from laboratory instrumentation involved in genetic testing. Because of the recent and ongoing development of these technologies (for example – Polymerase Chain Reaction and DNA Microarrays), standards are, at times, developing in an ad hoc manner. Some of this data is reported outside the logical boundaries of the laboratory itself and must be standardized to ensure interoperability with EHRs and PHRs.

- Need for standardization and harmonization of current textual interpretive report results in order to achieve interoperability.

Clinical Decision Support

Clinical decision support has the potential to advance the clinical practice of genetic/genomic testing and use of family health history information. However at the current time there are limited choices available and a lack of standardization in these systems. Some specific issues are as follows:

- Limited evidence and few clinical practice guidelines for use of genetic information.

- Fragmented informational databases without a standardized data structure which may be needed to provide clinical decision support capabilities.

ONC would like to receive feedback on the draft list of issues and obstacles and their descriptions for this use case. Please suggest additions, deletions and/or revisions.

5.0 Use Case Perspectives

The Personalized Healthcare Prototype Use Case will describe personalized healthcare from the viewpoint associated with three perspectives. The perspectives included in the use case are intended to indicate roles and functions, rather than organizations or physical locations. Each is described below:

- **Clinician**

  The clinician perspective includes family physicians, pediatricians, obstetricians, internists, nurse practitioners, physician assistants, genetic counselors, medical geneticists, and other personnel that conduct clinical assessment and management activities and participate in evaluation, diagnostic planning, genetic/genomic test ordering, and result interpretation activities.

- **Genetic Testing Laboratory**

  The genetic testing laboratory perspective includes medical laboratories which perform genetic laboratory tests ordered by clinicians to assess the genetic status of patients, generate data, and report results.

- **Consumer**

  The consumer perspective includes members of the public who receive healthcare services, as well as caregivers, patient advocates or surrogates, family members, and other parties who may be acting for, or in support of, a patient. The consumer self-reports family health history information, requests and views available family health history and genetic/genomic testing information, and considers personalized prevention messages and/or treatment information.

These perspectives are the focus of the events described in the candidate workflows.

ONC would like to receive feedback on the draft list of perspectives and their descriptions for this use case. Please suggest additions, deletions and/or revisions.

6.0 Candidate Workflows

The Personalized Healthcare Prototype Use Case focuses on the exchange of family health history and genetic/genomic testing information between consumers and clinicians in three candidate workflows.

6.1 Clinical Assessment

This candidate workflow is focused on gathering past medical history, current medical status, and family health history information from or by the consumer in an interoperable form to be used by clinicians. This information is accessed by clinicians for the purpose of developing a care plan.

- The clinician constructs a family health history or pedigree with entry of all medical problems. The clinician retrieves current patient family health history and any past genetic/genomic testing information from external sources to support the patient assessment.
    - Consumer self-reported family health history information may be available from the patient's PHR. Additional available information could be gathered electronically from hospital EHRs, ambulatory EHRs (such as from a Primary Care Physician (PCP)), family members' EHRs, and other sources that hold information about the patient.
    - The clinician and support staff may also gather family health history information by interviewing the patient, patient's family, significant others, and/or caregivers – and in some instances, by contacting the patient's PCP.
- The clinician performs interpretation, assembly, validation, and evaluation activities with support from clinical decision support tools. The clinician analyzes the relevant medical and health information for genetic and/or birth defect risks and assesses and interprets the risk for occurrence (or recurrence) of genetic conditions. The clinician may select, validate, and incorporate family health history information into the EHR.
    - The clinical decision support tools could utilize data tables, risk assessment algorithms, and other information from the genetic/genomic knowledge repository.
    - The process of setting up clinical decision support relies on standard vocabularies for family relationship nomenclature and other critical items related to family health history, and standards for the communications between the EHR knowledge source(s) and application(s).

- If appropriate, the clinician will order additional genetic/genomic tests.
    - Additional genetic/genomic tests for the patient may be ordered through a web application or through an available EHR.
    - The order may be made utilizing clinical decision support tools, and may include accompanying reference information such as patient information to be considered in analysis and interpretation of the results, general specimen information, billing information, and physician and patient contact information.
    - In the special case of mandated newborn genetic screening, a delivering clinician or other healthcare provider will order some number of genetic tests. The specific tests mandated vary from state to state but the information flow may be handled in the same manner.

ONC would like to receive feedback regarding the information needs associated with mandated newborn genetic screening and how they differ from other genetic tests.

6.2 Genetic Testing and Reporting

This candidate workflow is focused on a genetic testing laboratory receiving and capturing genetic/genomic test orders and any accompanying information necessary for the testing, as well as the ability to exchange genetic/genomic laboratory test results among laboratories and ordering clinicians with appropriate privacy and security considerations.

- The genetic testing laboratory receives and captures the genetic/genomic testing orders and accompanying information necessary for the testing.
    - Because of the specialized nature of genetic testing and the evolving technologies, situations may arise in which the testing lab will need to communicate back to the ordering clinician to ensure that the correct testing has been ordered and all the necessary information has been gathered to enable testing to take place. This information can be exchanged directly through EHRs and could potentially utilize clinical decision support.
- The genetic testing laboratory performs the technical steps required to produce the genetic/genomic data.
- The genetic testing laboratory develops the patient report and transmits it to the ordering clinician's EHR or other clinical data system. Genetic/genomic knowledge repositories may be utilized by the genetic testing laboratory to support this activity.

- The ordering clinician receives results from the genetic testing lab.
- Results (de-identified or appropriately consented) may also be used to populate databases of clinical information for research purposes.

ONC would like to receive feedback on the information needs associated with a clinician requesting additional test information from the genetic testing laboratory. Are there standardization needs for the types of information that need to be exchanged in this regard? ONC would also like to receive feedback regarding the needs around information being accessed from instrumentation in a standardized format suitable for the clinician EHRs. Is this an issue that should be specifically addressed within the draft detailed use case?

6.3 Clinical Management

This candidate workflow is focused on determining appropriate preventative action, treatment protocol, messaging, and interpretation of results utilizing clinical decision support tools, and genetic/genomic knowledge repositories, as well as the consumer's ability to permit designated individuals to request and view information in their PHR. Additionally, this candidate workflow highlights the consumer's ability to consider personalized prevention messages received via their PHR.

- The ordering clinician receives results from the genetic testing lab.
    - The ordering clinician performs interpretation, care planning and care plan implementation activities utilizing clinical decision support, and communicates this information to the patient.
    - The ordering clinician provides results and additional interpretation from the genetic testing lab to other clinicians, such as the next provider of care (if applicable), and the consumer. This information can be incorporated into clinicians' EHRs and into the patient's PHR.
- The consumer receives testing results and care plan from the communicating clinician or their PCP.
- The consumer requests available family health history and genetic/genomic testing information via their PHR.
    - This may have been previously 'pushed' to the consumer.
- The consumer permits designated individuals to request and view information in their PHR (a.k.a., proxy access).

  - The consumer's family members should have the ability to view, select, and incorporate information into their own PHRs.

- The consumer considers personalized prevention messages received via their PHR.

ONC would like to receive feedback on the personalized healthcare use case candidate workflows. Should any changes be made to the descriptions of these interactions? For those candidate workflows listed, is the working definition of key information sources and recipients sufficient? If not, what changes should be made?

Appendix A: Glossary

AHIC: American Health Information Community.

Care: Relieving the suffering of individuals, families, communities, and populations by providing, protecting, promoting, and advocating the optimization of health and abilities.

CCHIT: Certification Commission for Healthcare Information Technology.

Clinicians: Healthcare providers with patient care responsibilities, including physicians, advanced practice nurses, pharmacists, physician assistants, nurses, medical geneticists, genetic counselors, and other credentialed personnel involved in treating patients.

Consumers: Members of the public who may receive healthcare services. These individuals may include: caregivers, patient advocates, surrogates, family members, and other parties who may be acting for, or in support of, a patient.

Department of Health and Human Services (HHS): This is the federal agency responsible for human health, and has oversight over many other federal agencies such as the Food and Drug Administration (FDA), the National Institutes of Health (NIH), the Centers for Disease Control and Prevention (CDC), the Centers for Medicare and Medicaid Services (CMS), the Agency for Health Research and Quality (AHRQ), the Substance Abuse and Mental Health Services Administration (SAMHSA), and others.

Electronic Health Record (EHR): The electronic health record is a longitudinal electronic record of patient health information generated in one or more encounters in any care delivery setting. This information may include patient demographics, progress notes, problems, medications, vital signs, past medical history, immunizations, laboratory information and radiology reports.

Genetic/Genomic Knowledge Repositories: Organizations that maintain resources, such as online servers and databases, which provide evidence-based analysis and validated knowledge, such as information on specific genetic diseases, human genomic sequence data and references to the relevant medical literature.

Genetic Testing Laboratories: Medical laboratories which perform genetic laboratory tests ordered by genetics specialists to assess the genetic status of patients. These services may be delivered through hospitals or through free-standing entities.

Health Information Exchange (HIE): A multi-stakeholder entity that enables the movement of health-related data within state, regional, or non-jurisdictional participant groups.

Health Researchers: Organizations or individuals who use health information to conduct research.

Healthcare Entities: Organizations that are engaged in or support the delivery of healthcare. These organizations could include hospitals, ambulatory clinics, long-term care facilities, community-based healthcare organizations, employers/occupational health, school health, dental clinics, psychology clinics, care delivery organizations, and other healthcare facilities.

Healthcare Payors: Insurers, including health plans, self-insured employer plans, and third party administrators, providing healthcare benefits to enrolled members and reimbursing provider organizations. As part of this role, they provide information on eligibility and coverage for individual consumers, as well as claims-based information on consumer medication history. Case management or disease management may also be supported.

HITSP: Healthcare Information Technology Standards Panel.

ONC: Office of the National Coordinator for Health Information Technology.

Patients: Members of the public who receive healthcare services.

Personal Health Record (PHR): A health record that can be created, reviewed, annotated, and maintained by the patient or the caregiver for a patient. The personal health record may include any aspect(s) of the health condition, medications, medical problems, allergies, vaccination history, visit history, or communications with healthcare providers.

Point-to-Point Exchange: A direct link or communication connection with defined endpoints.

Public Health Agencies (local/state/federal): Local, state, and federal government organizations and personnel that exist to help protect and improve the health of their respective constituents.

Registries: Organized systems for the collection, storage, retrieval, analysis, and dissemination of information on individual persons to support health needs.